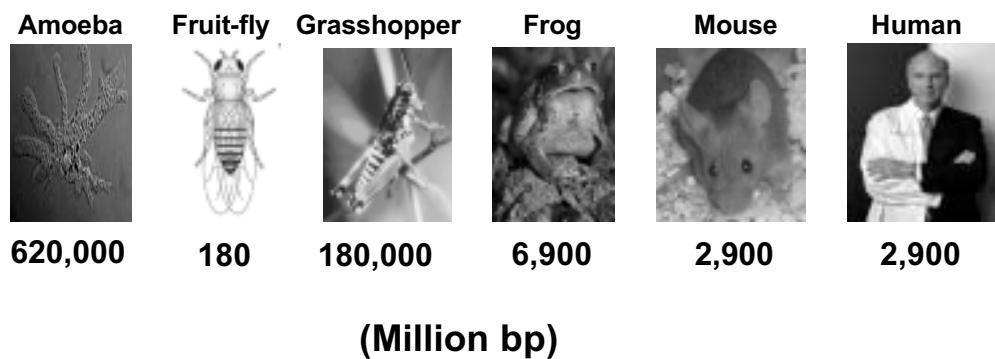


- **2-5% of DNA is translated**
- **25-30% of DNA is transcribed**
- **60-70% of the genome contains non-genic DNA**

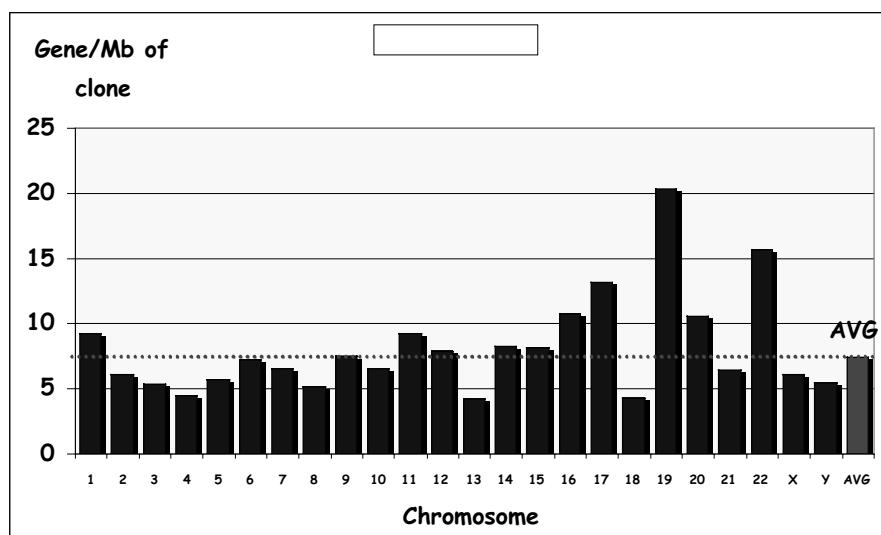
Why do we have a 2,900,000,000 bp Genome?

C-value Paradox

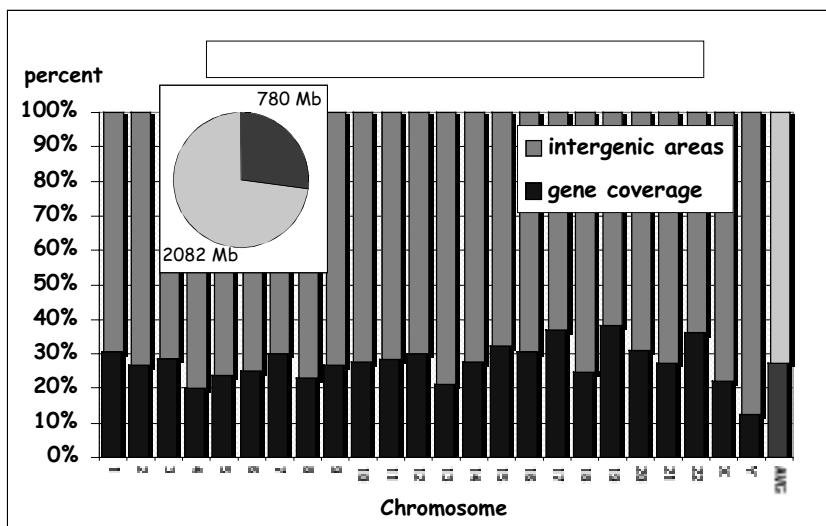
Genome size does not correlate with organismal complexity



Gene Density in the Human Genome



Gene Coverage of the Human Genome



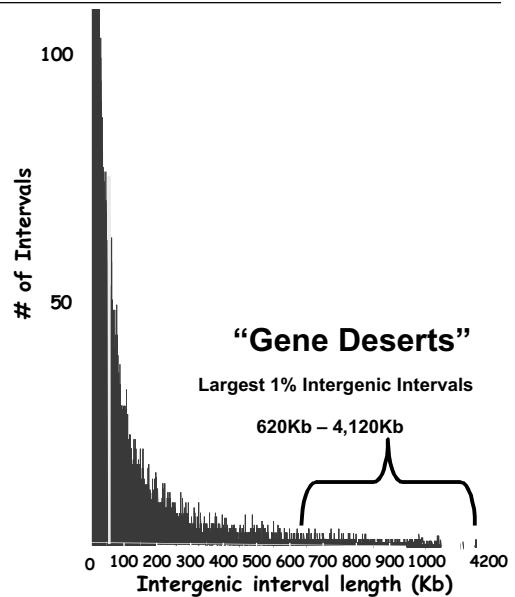
Defining “Gene Deserts”

1 – Calculate Intergenic Lengths

EMSEMBL- 21,978 genes

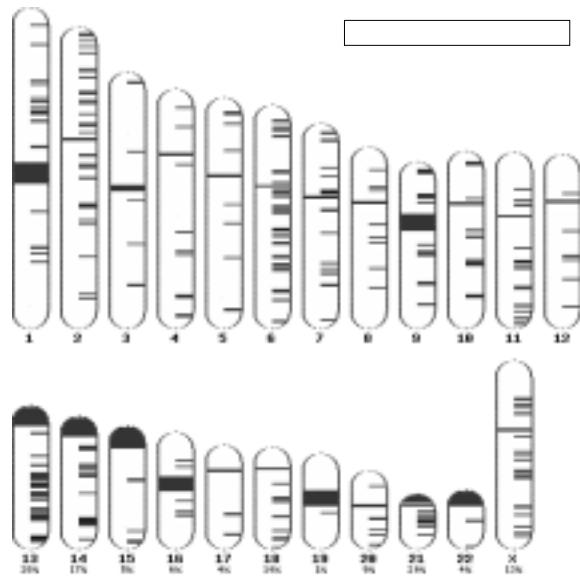
REFSEQ Annotation- 12,439 genes

2 – Exclude Heterochromatic DNA & Clone Gaps.

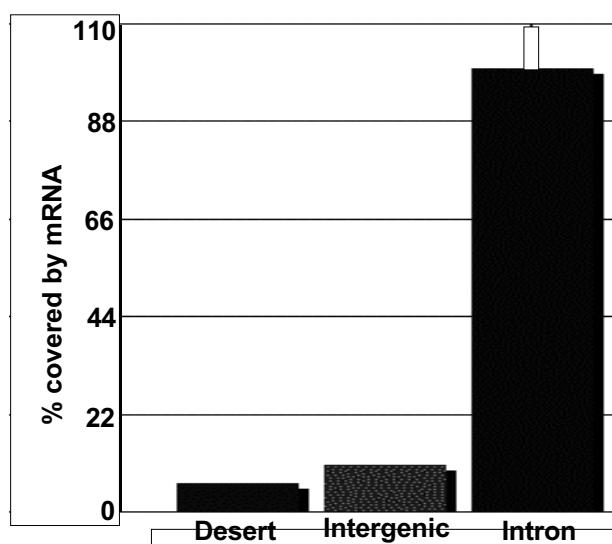


Distribution of Human “Gene Deserts”

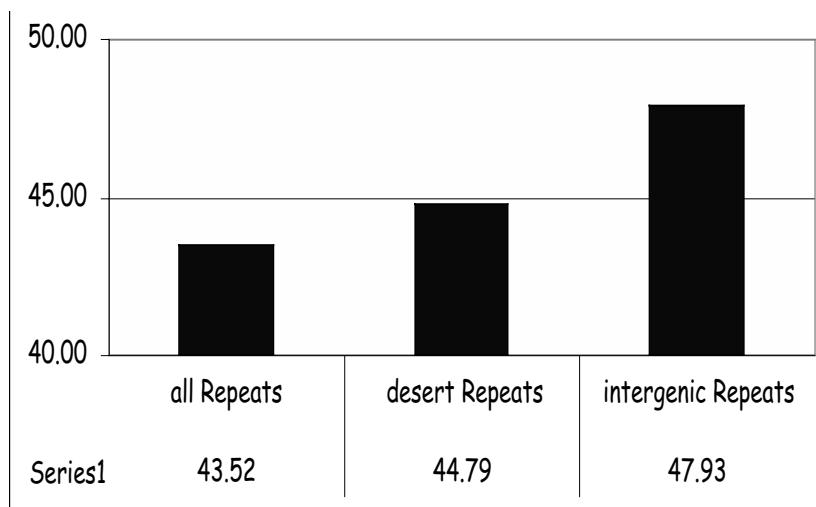
- Total #
 - 234 deserts
- Size Range
 - 680 – 4,120 Kbp
- % of Genome
 - 9% (277 Mbp)



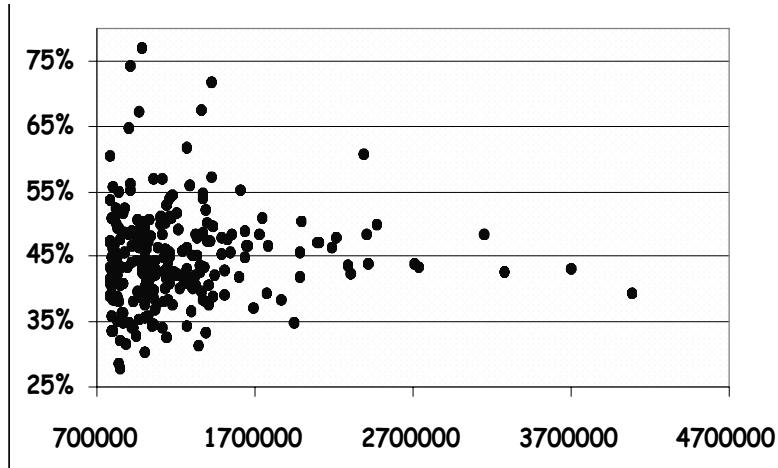
mRNA Content of Genomic Regions



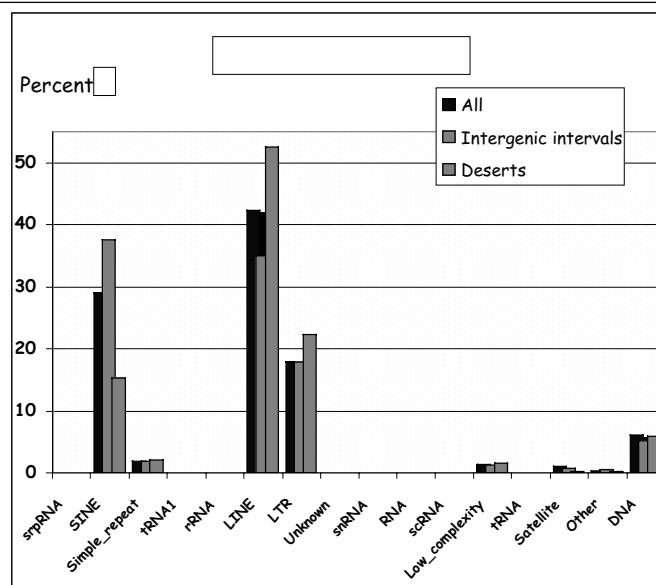
Distribution of Repetitive Elements in the Genome



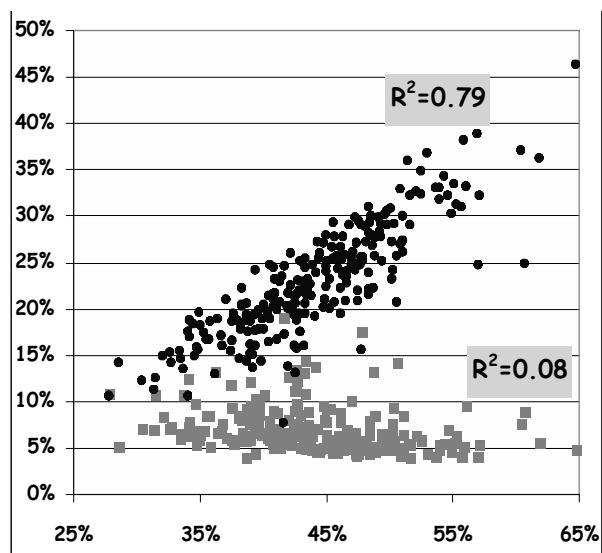
Repeat Content and Length of Gene Deserts



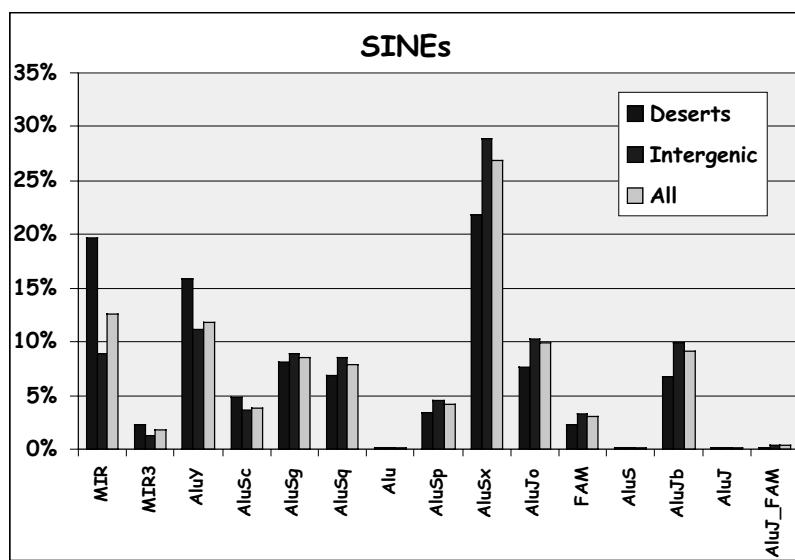
Distribution of Repetitive Elements in the Genome



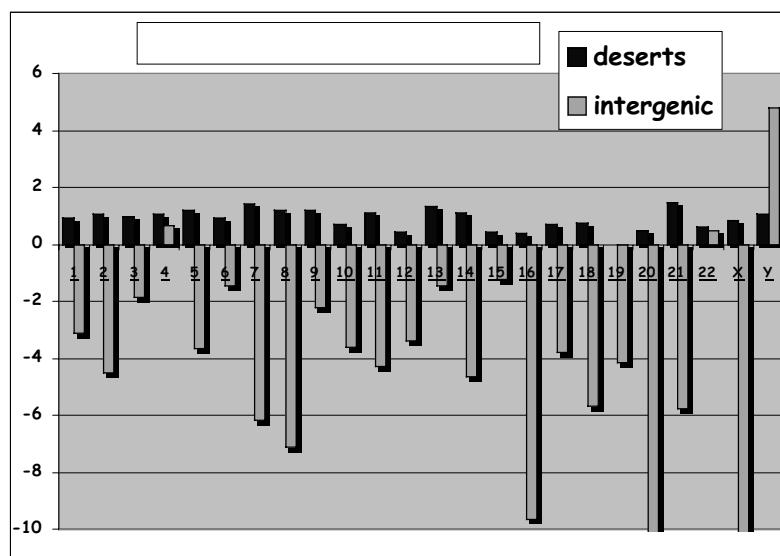
SINE and LINE vs Repeat Content



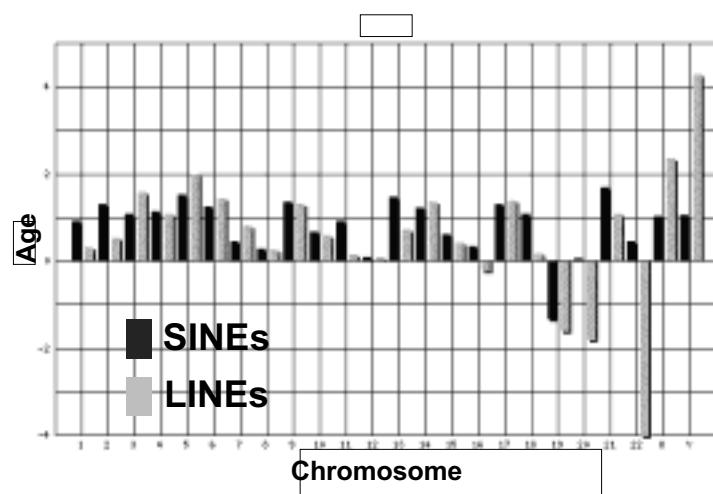
Age of SINE Elements in Intergenic DNA



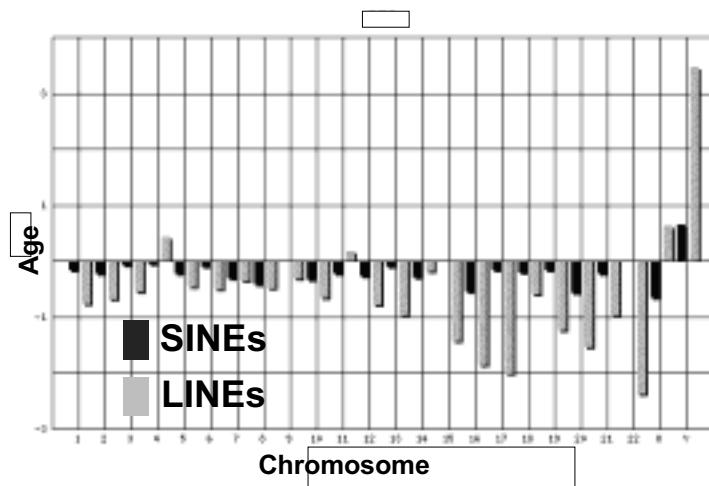
Age of SINES in Intergenic DNA



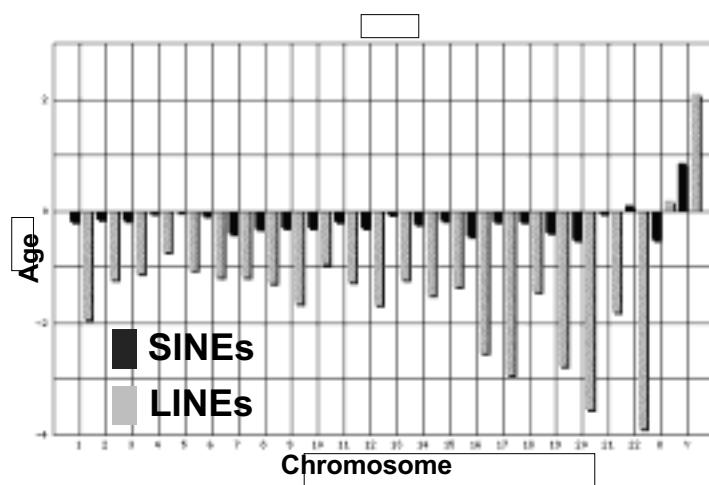
Age of Repetitive Elements in Gene Deserts



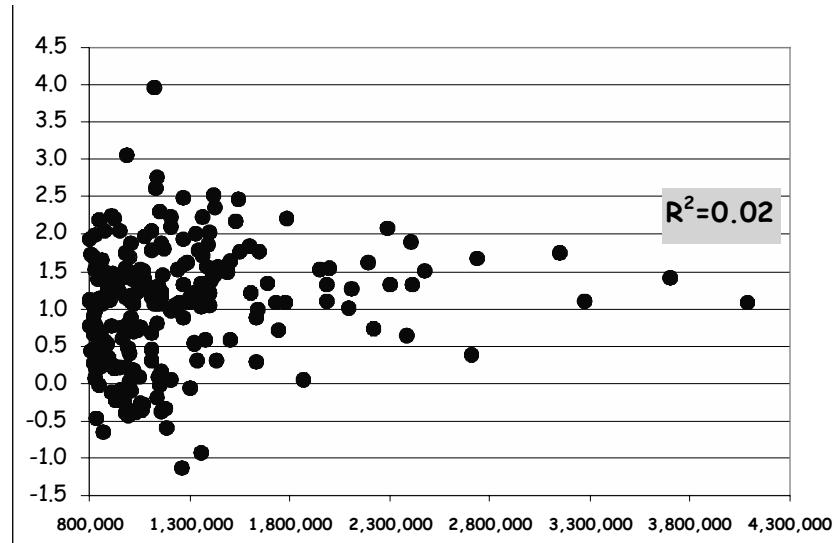
Age of Repetitive Elements in Intergenic Regions



Age of Repetitive Elements in Introns



Age of repetitive Elements vs Length of Gene Deserts



Summary of Human Gene Deserts Findings

- 1- Don't appear to result from the expansion of genomic segments due to transposon insertions**
- 2 – Contain primarily younger repetitive elements than the intergenic or intronic fraction of the genome**
- 3 – The amount of younger insertions doesn't predict length of the gene desert**

Are gene deserts artifacts of genomic annotation?

Can the younger age of insertions reflect a higher degree of “housekeeping” deletions in deserts?

Are Human “Gene Deserts” Also Deserts in Mouse?

Strategy for Identifying Mouse Gene Deserts

1 –BLAT Comparison of Human “Gene Deserts” to Mouse Genome Assembly

2 – Search for predicted genes in orthologous mouse DNA:

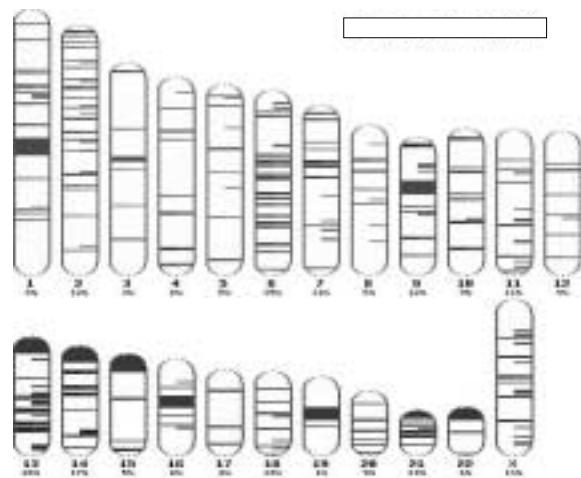
-Public Mouse Assembly
RefSeq Annotation(8,285 genes)

-Celera Mouse Assembly
Less than 2 lines of evidence for gene prediction

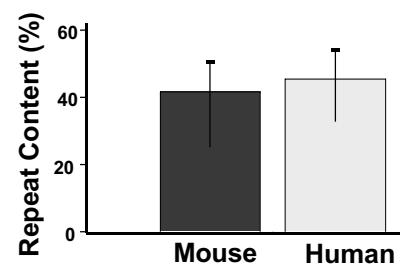
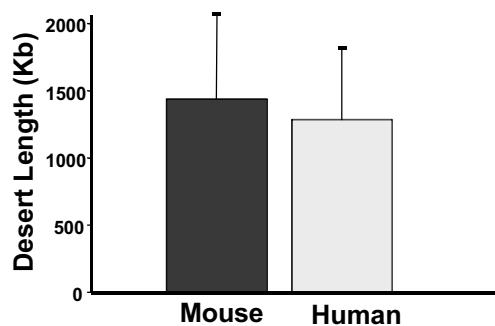
Orthologous Mouse “Gene Deserts”

- HUMAN
 - 234 Gene Deserts

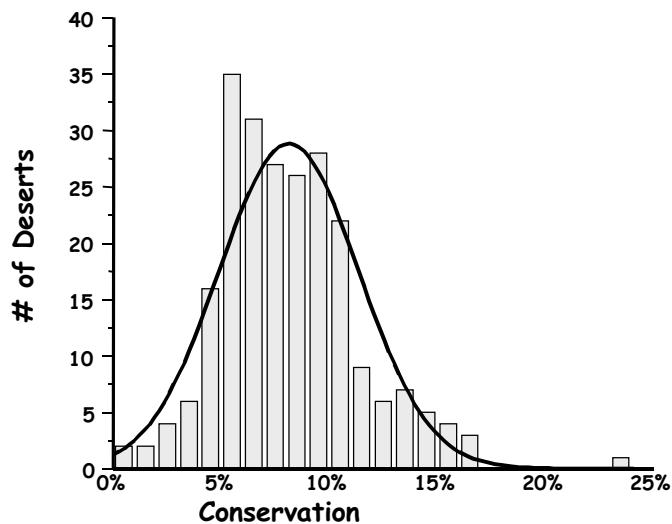
- Orthologous Mouse Comparison
 - 178 (74%) are also Deserts



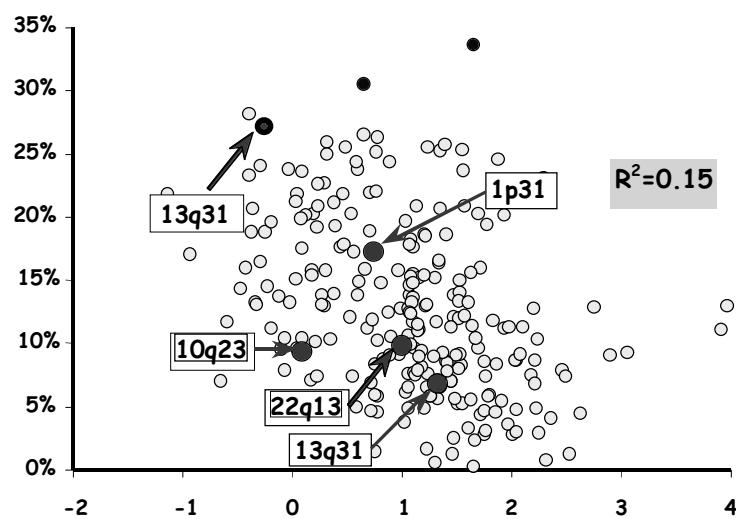
Human/Mouse “Gene Deserts” Comparisons



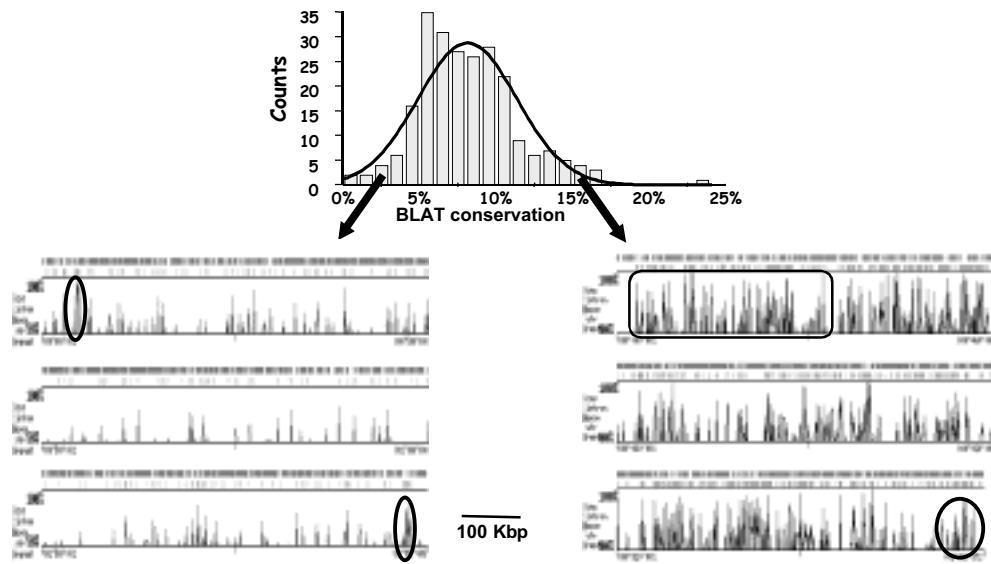
Human – Mouse Conservation of “Gene Deserts”



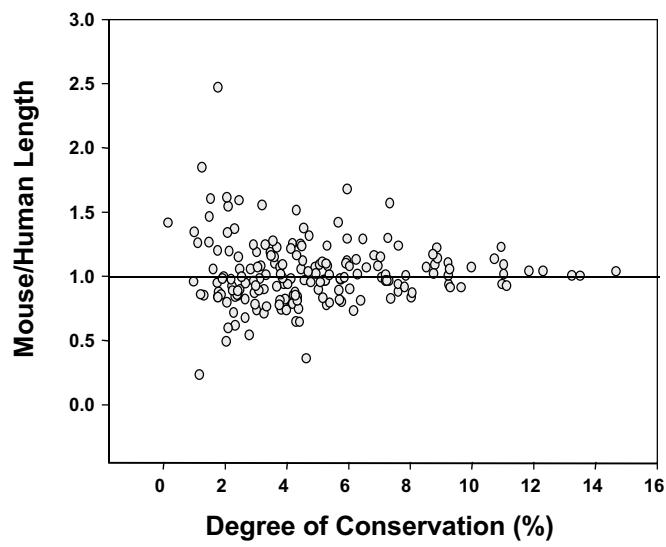
Human-mouse Conservation and Age of Gene Deserts



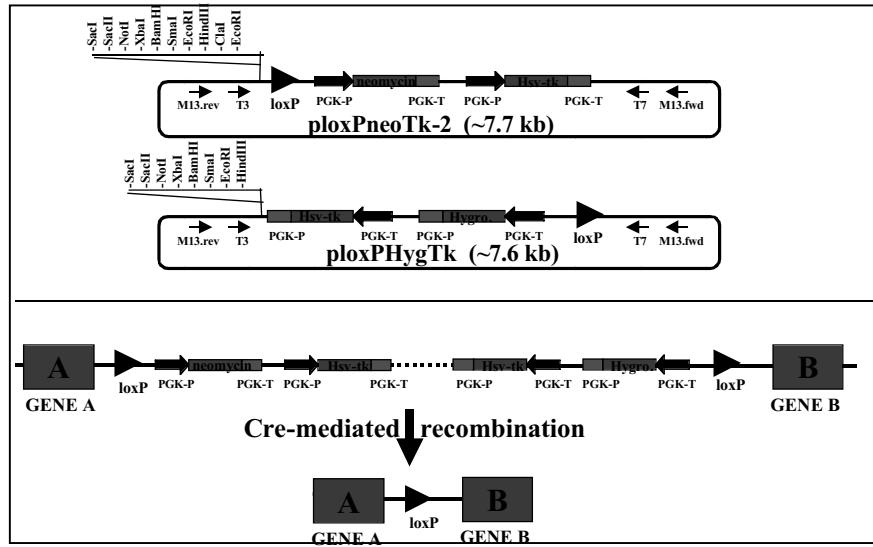
Human-Mouse Conservation in Gene Deserts



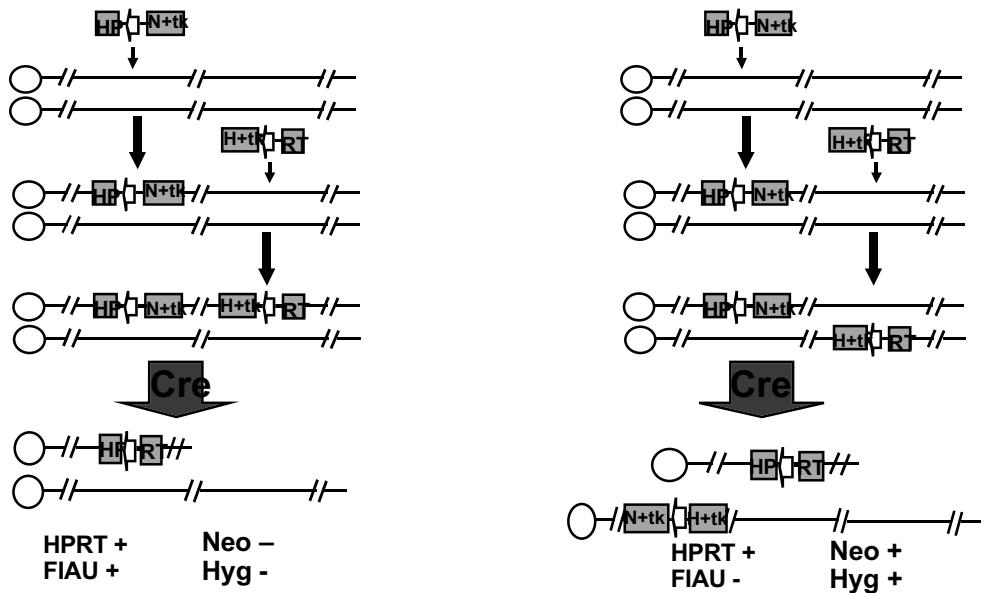
Conservation of Sequence and Length of Gene Deserts



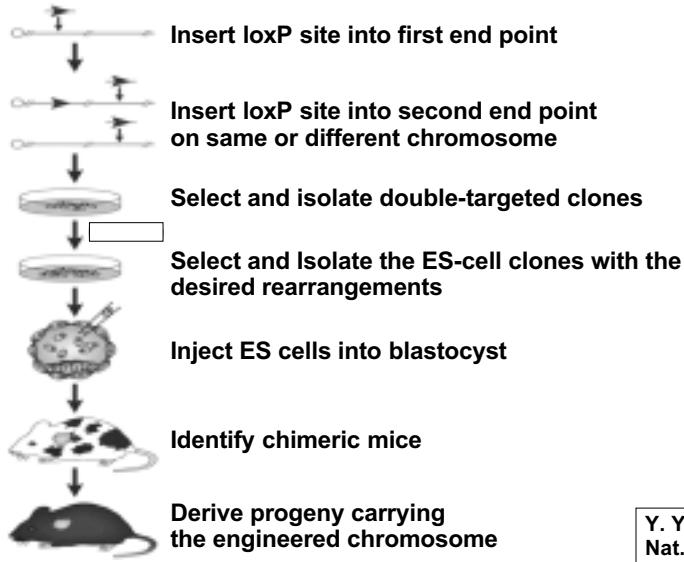
Cre-loxP-mediated Chromosomal Rearrangement



Cis and Trans Chromosomal Targetting



Generation of Chromosomal Deletions in Mice



Y. Yu and A. Bradley
Nat. Rev. Gen. 2, 780 -790 (2001)

Determining function of “Gene Deserts”

